NAGS gene

N-acetylglutamate synthase

Normal Function

The *NAGS* gene provides instructions for making the enzyme N-acetylglutamate synthase. This enzyme is needed for the urea cycle, a series of reactions that occurs in liver cells. The urea cycle processes excess nitrogen, generated when protein is used by the body, into a compound called urea that is excreted by the kidneys. Excreting the excess nitrogen prevents it from accumulating in the form of ammonia, which is toxic, especially to the nervous system.

N-acetylglutamate synthase controls the production of a compound called N-acetylglutamate in the mitochondria, the energy-producing centers in cells. N-acetylglutamate is necessary to activate the enzyme carbamoyl phosphate synthetase I. This enzyme controls the first step of the urea cycle, in which excess nitrogen compounds are incorporated into the cycle to be processed.

Health Conditions Related to Genetic Changes

N-acetylglutamate synthase deficiency

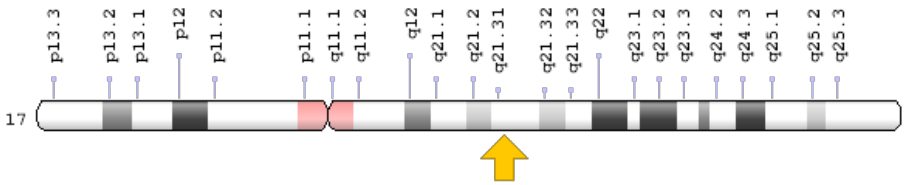
Approximately 12 *NAGS* gene mutations have been identified in people with N-acetylglutamate synthase deficiency. A mutated *NAGS* gene may result in an N-acetylglutamate synthase enzyme that is shorter than normal or the wrong shape, or may prevent the enzyme from being produced at all.

The shape of an enzyme affects its ability to control a chemical reaction. If the N-acetylglutamate synthase enzyme is damaged or missing, N-acetylglutamate will be produced in lower-than-normal amounts, or not at all. This shortage of N-acetylglutamate interferes with proper activation of carbamoyl phosphate synthetase I, preventing it from fulfilling its role in the urea cycle. Excess nitrogen is not converted to urea for excretion, and ammonia accumulates in the body. This accumulation causes neurological problems and other signs and symptoms of N-acetylglutamate synthase deficiency.

Chromosomal Location

Cytogenetic Location: 17q21.31, which is the long (q) arm of chromosome 17 at position 21.31

Molecular Location: base pairs 44,004,546 to 44,009,068 on chromosome 17 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- AGAS
- ARGA
- MGC133025
- NAGS_HUMAN
- NAT7

Additional Information & Resources

Educational Resources

- Biochemistry (5th editions, 2002): Ammonium Ion is Converted into Urea in Most Terrestrial Vertebrates
  https://www.ncbi.nlm.nih.gov/books/NBK22450/

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28NAGS%5BTIAB%5D%29+OR+%28N-acetylglutamate+synthase%5BTIAB%5D%29%29+OR+%28%28AGAS%5BTIAB%5D%29+OR+%28ARGA%5BTIAB%5D%29+OR+%28MGC133025%5BTIAB%5D%29+OR+%28NAT7%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- N-ACETYLGLUTAMATE SYNTHASE
  http://omim.org/entry/608300

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=NAGS%5Bgene%5D
- HGNC Gene Family: GCN5 related N-acetyltransferases
  http://www.genenames.org/cgi-bin/genefamilies/set/1134
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=17996
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/162417
- UniProt
  http://www.uniprot.org/uniprot/Q8N159

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Reprinted from Genetics Home Reference:
https://ghr.nlm.nih.gov/gene/NAGS

Reviewed: October 2006
Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
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